THE SCIENCE OF HEREDITY AND VARIATION

Lecture 1

GENETICS

- the science of heredity and variation

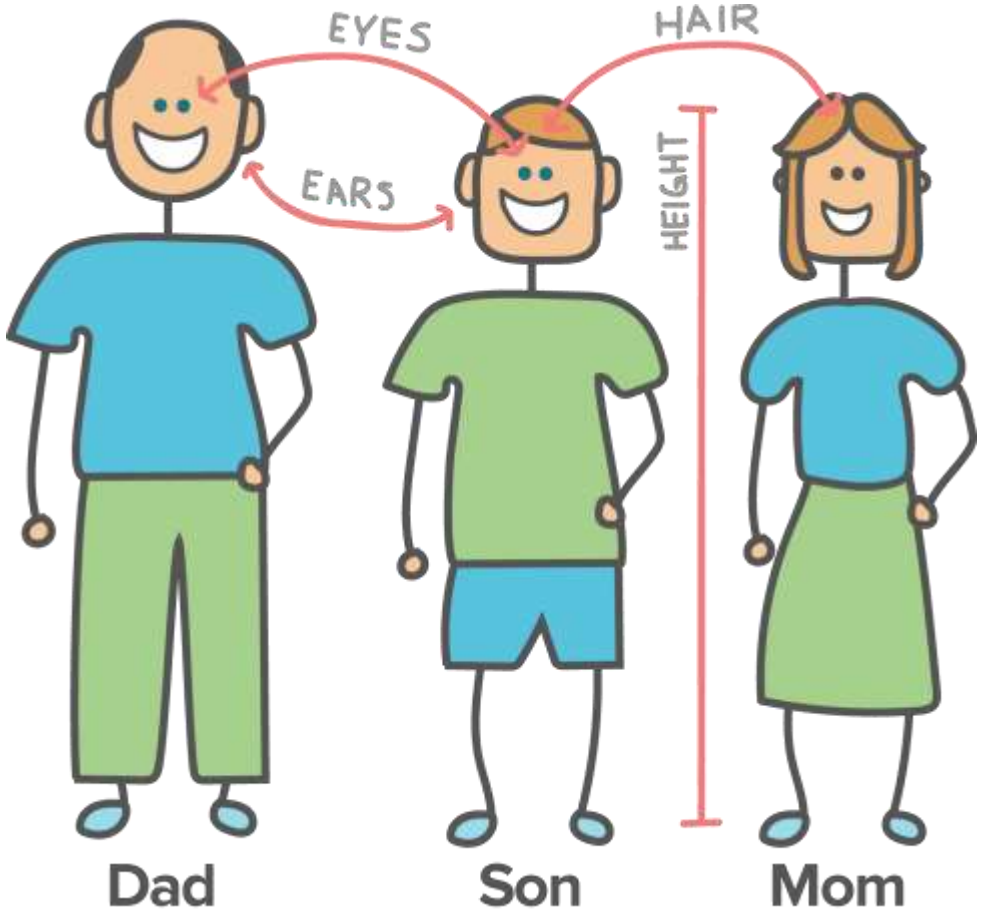
- Etymology
  - Greek word "*gen*"
    - to become or to grow into something
    - coined by William Bateson in 1906

Heredity

- transmission of traits from parents to offsprings
- similarity of parents and offspring

Variation

- differences between parents and offsprings
- differences among offsprings
- differences among individuals



heredity

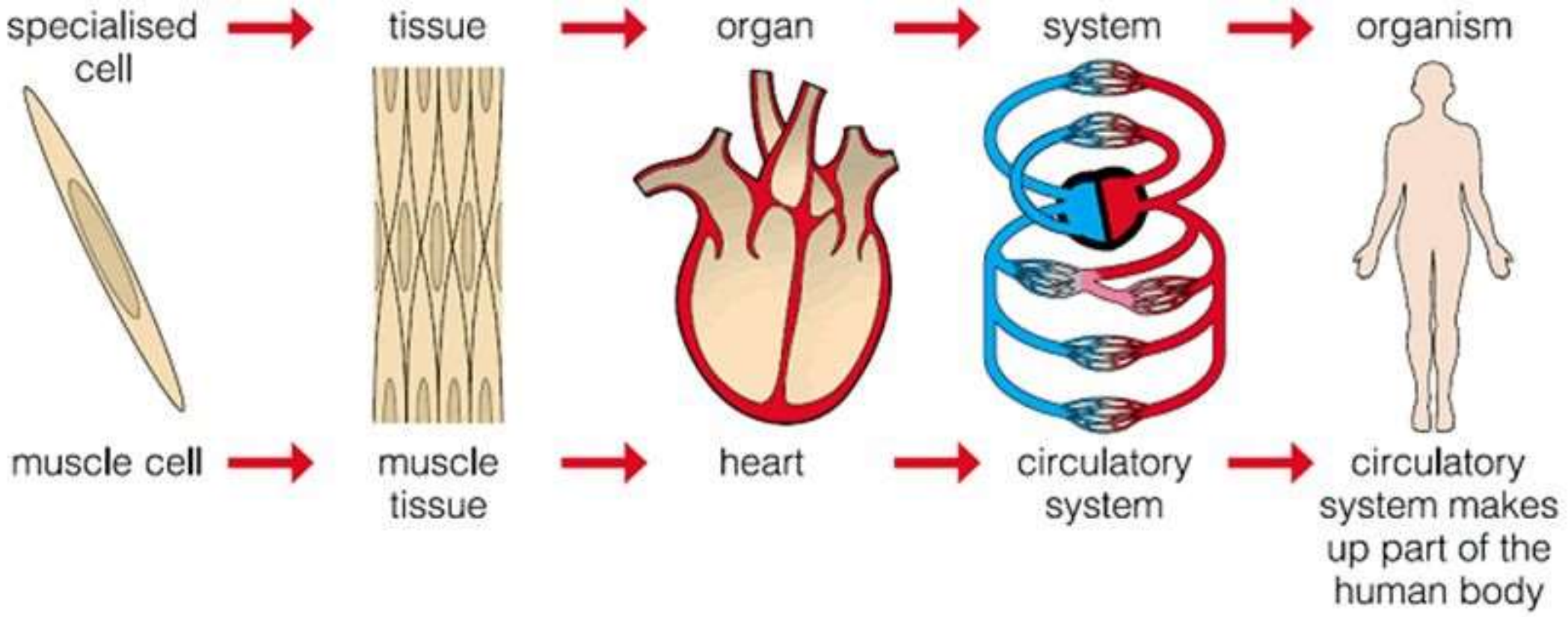
variation

“Genetics is the most interesting and the most fascinating field in biology”

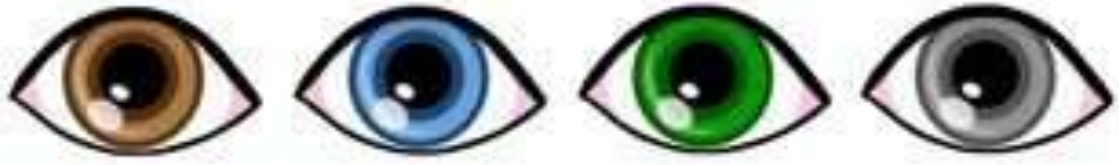
Importance???

"Genes are the principal determinants of life processes."

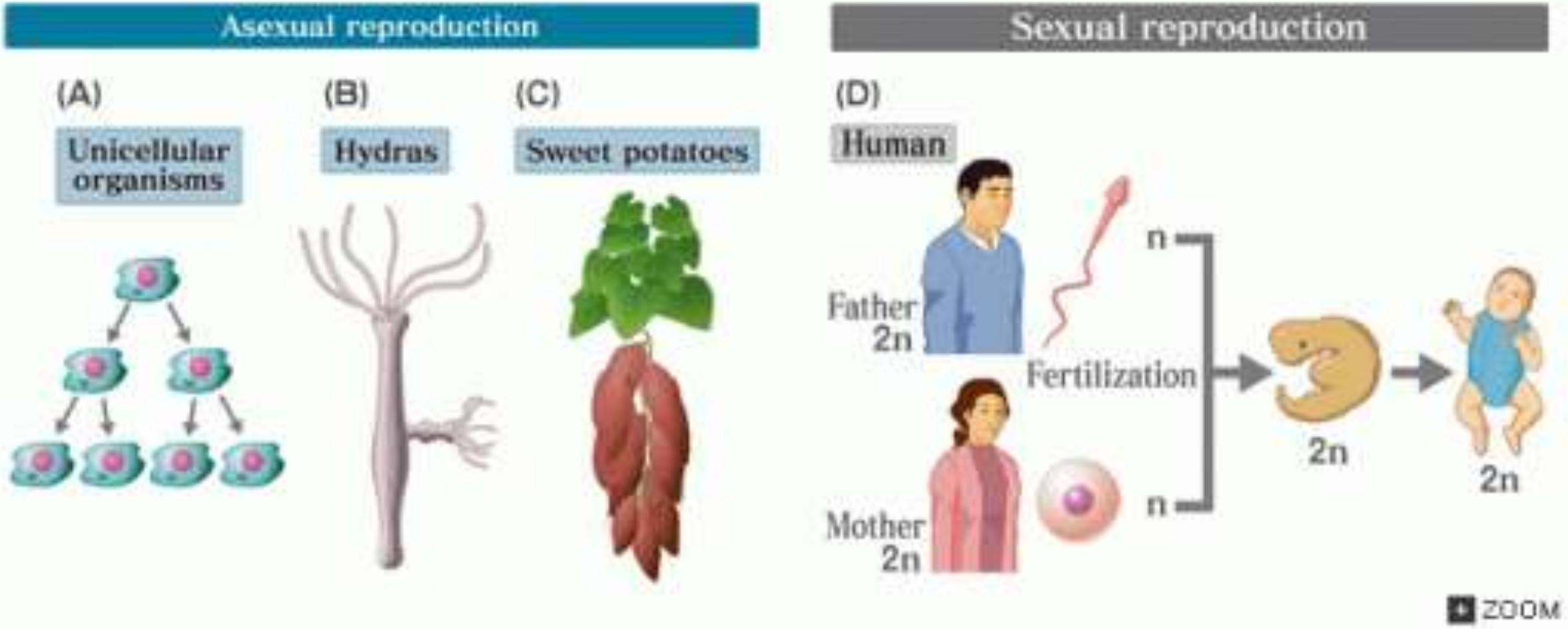
- organization of cells into tissues/organs
- organism appearance
- reproduction
- adaptability
- behaviour



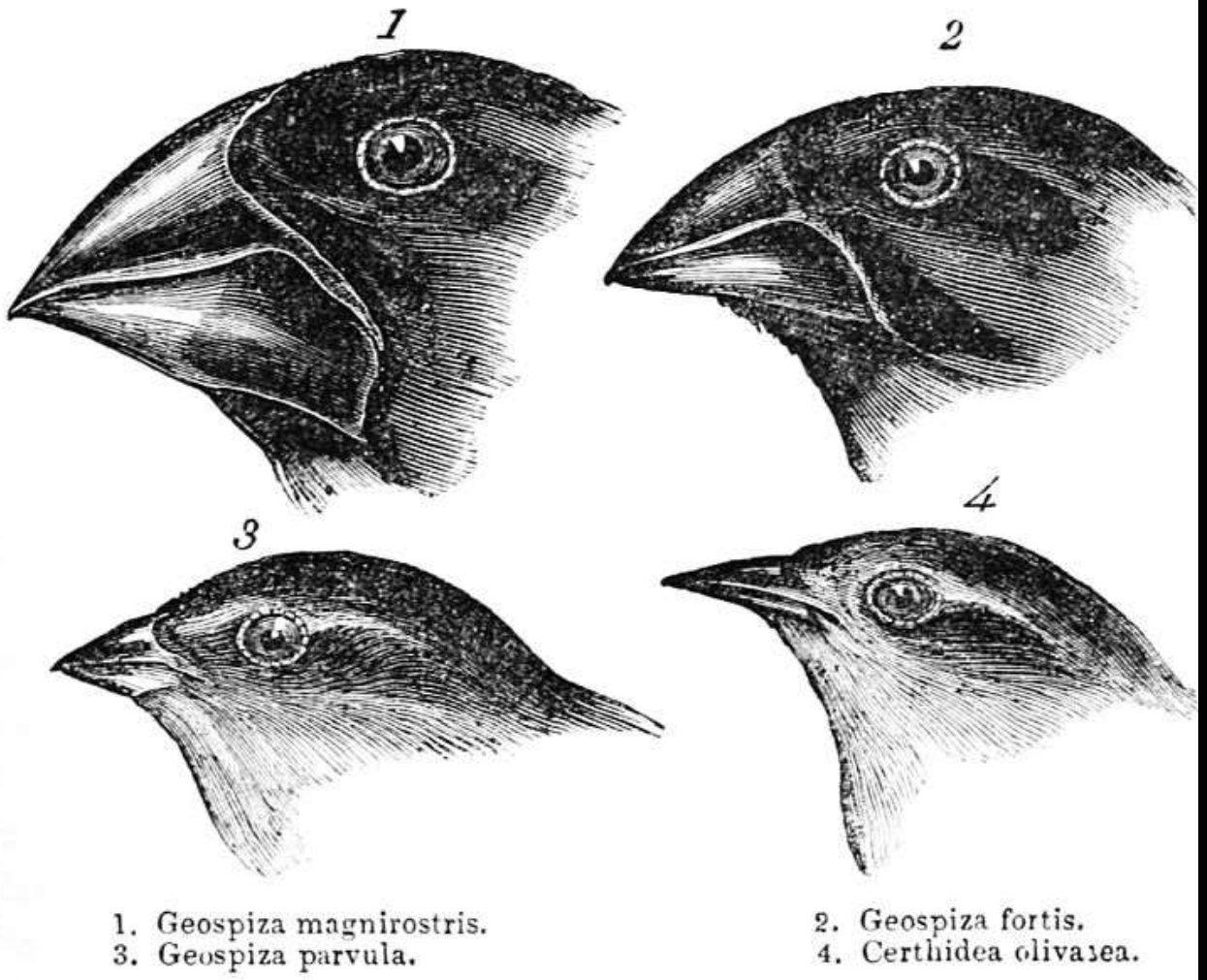
organization of cells into tissues/organs

Gene	Alternative Alleles
Eye colour	 Brown Blue Emerald Grey
Hair colour	Blonde Red Brown Black

organism appearance



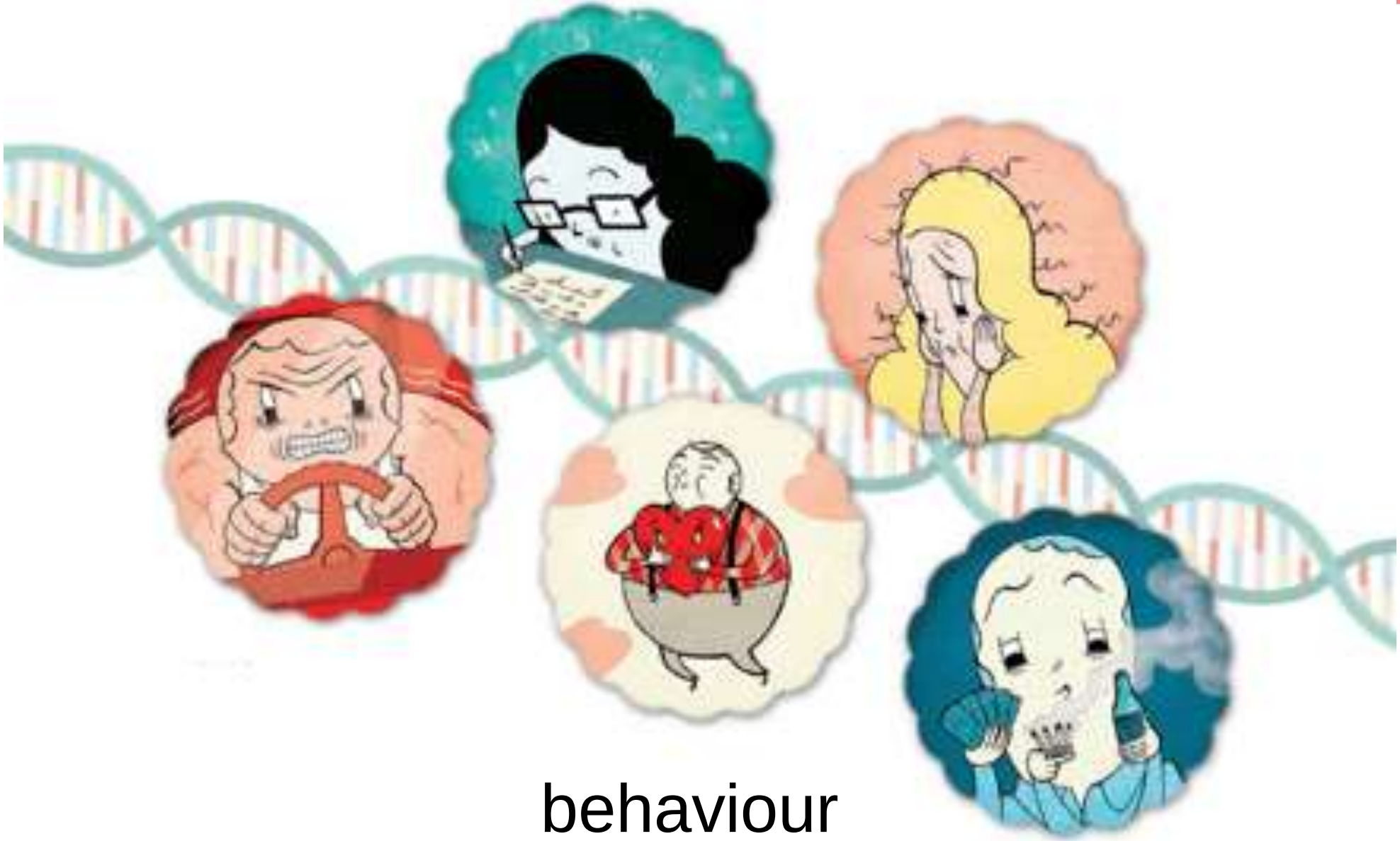
reproduction



1. Geospiza magnirostris.
3. Geospiza parvula.

2. Geospiza fortis.
4. Certhidea olivacea.

adaptability



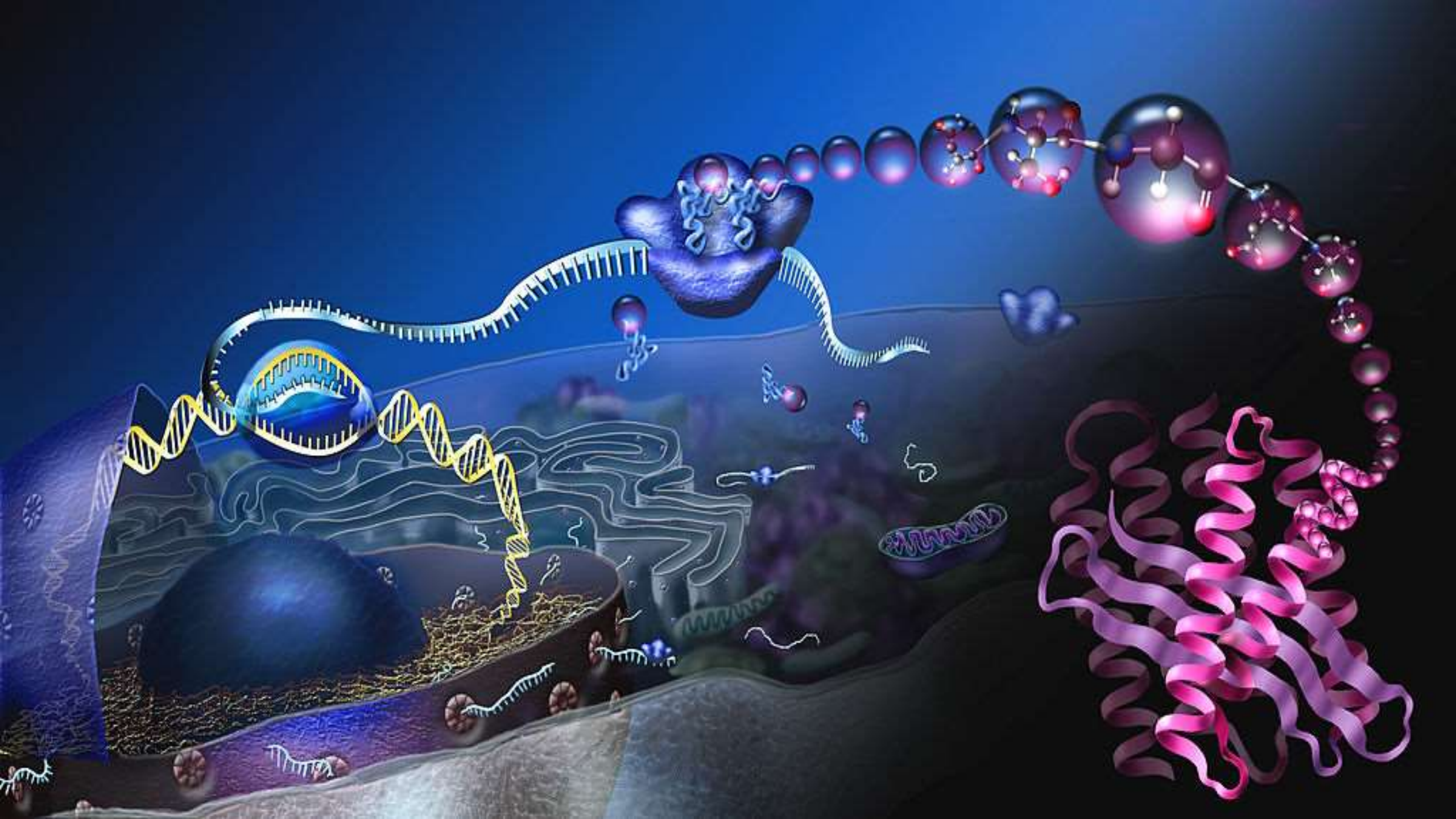
behaviour

BRANCHES of GENETICS

1. Molecular Genetics
2. Developmental Genetics
3. Cytogenetics
4. Evolutionary Genetics
5. Biochemical Genetics
6. Behavioral Genetics
7. Population Genetics
8. Quantitative Genetics

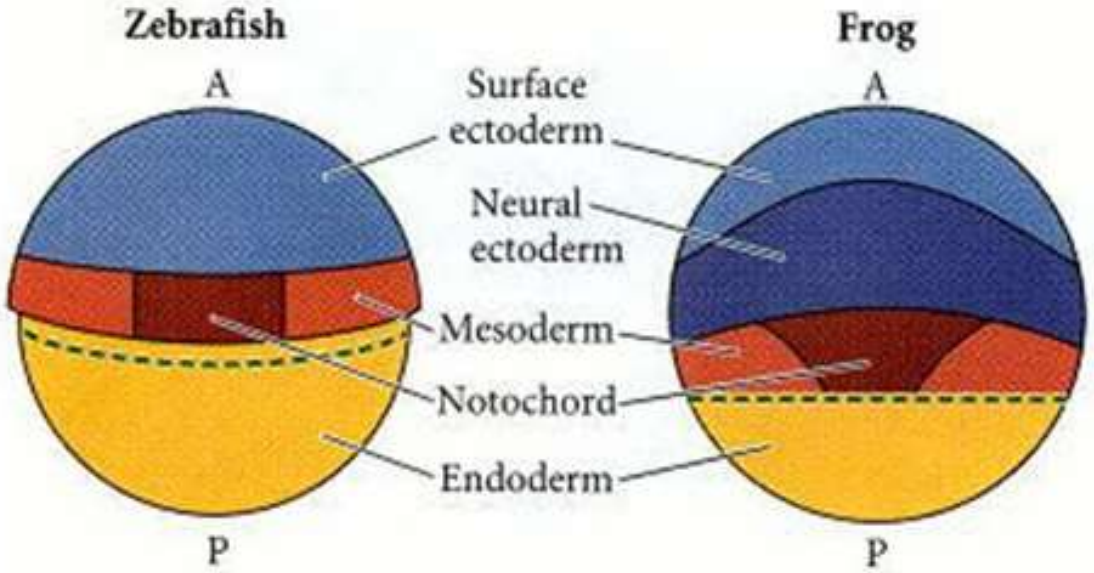
BRANCHES of GENETICS

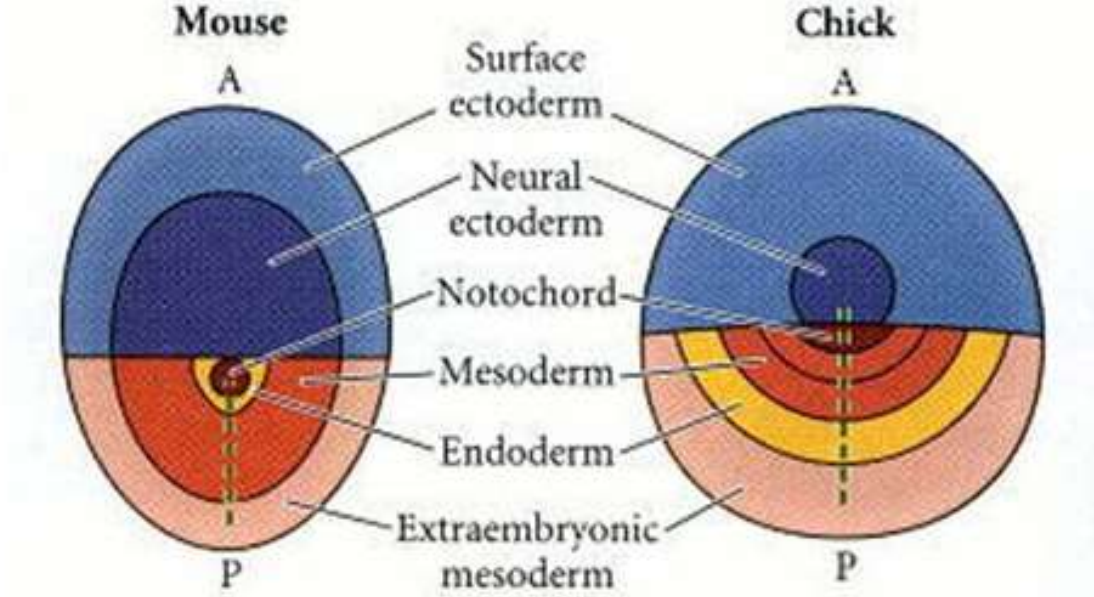
1. Molecular Genetics
   - structure and functions of genes at the molecular level
   - Central Dogma of Molecular Biology
     - replication
     - transcription
     - translation



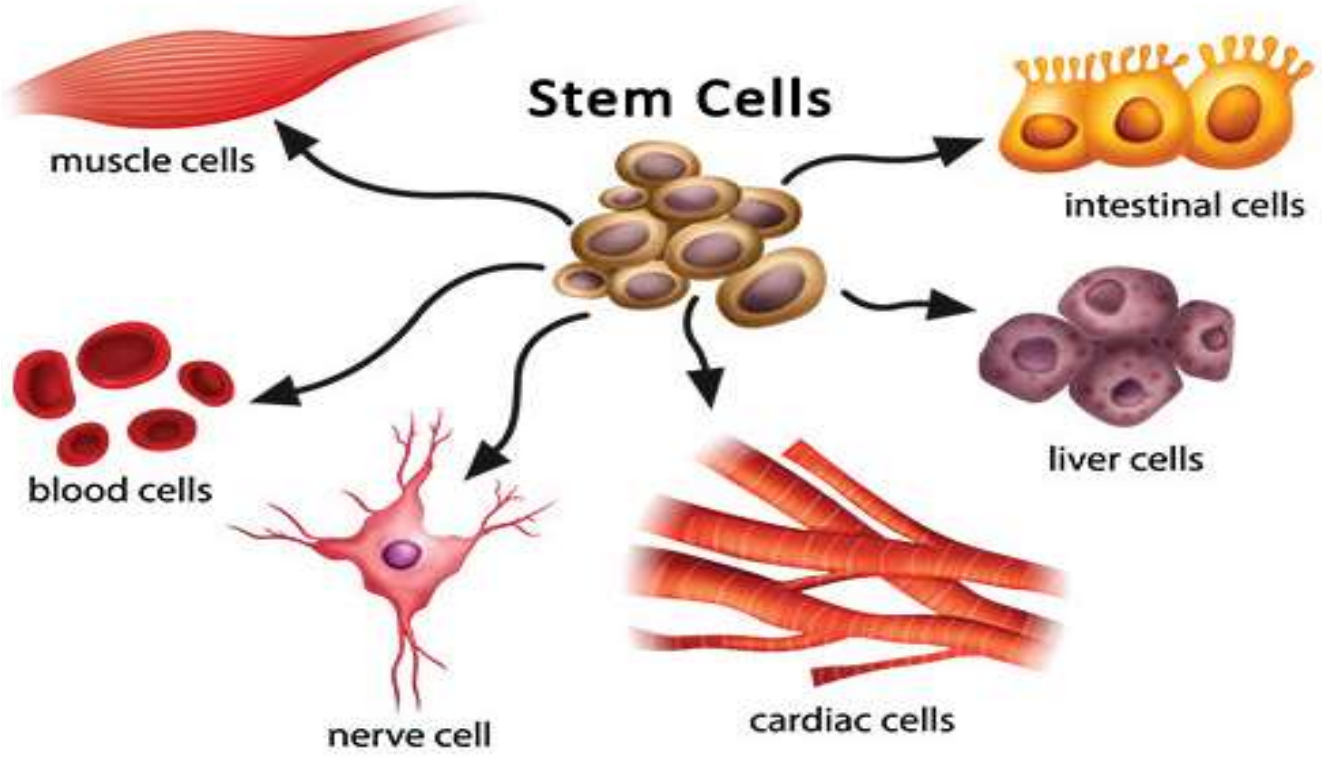
BRANCHES of GENETICS

2. Developmental Genetics
   - gene regulation during development
   - switching on and off of genes



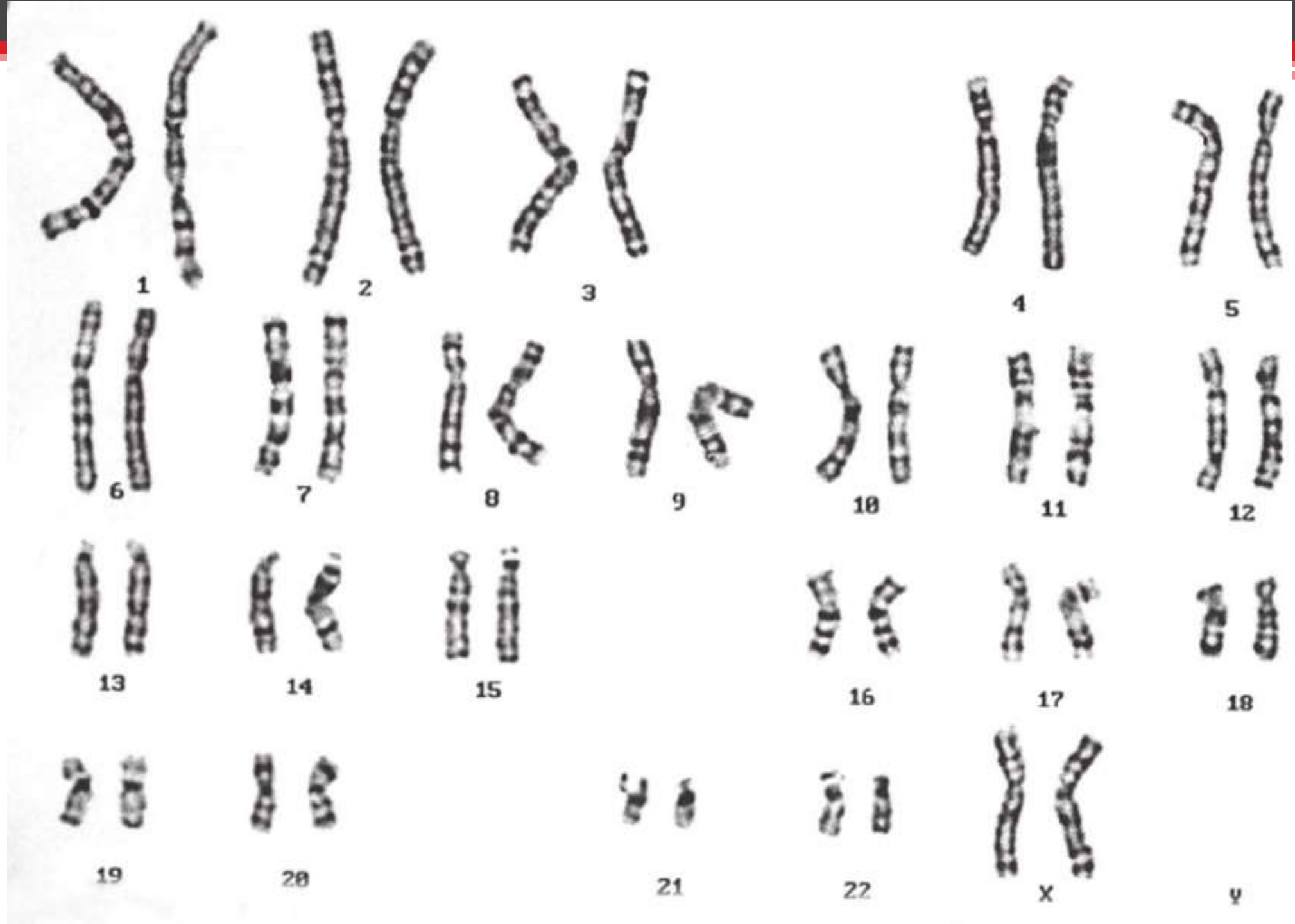


- differentiation of cells requires regulation of specific genes
- timing of gene regulation can determine when a tissue develops or when cells undergo apoptosis



BRANCHES of GENETICS

3. Cytogenetics
   - behavior of chromosomes
   - carriers of genes
   - e.g. Down's Syndrome (extra chromosome 21)
     - 22IIA + I^{21} , XX or XY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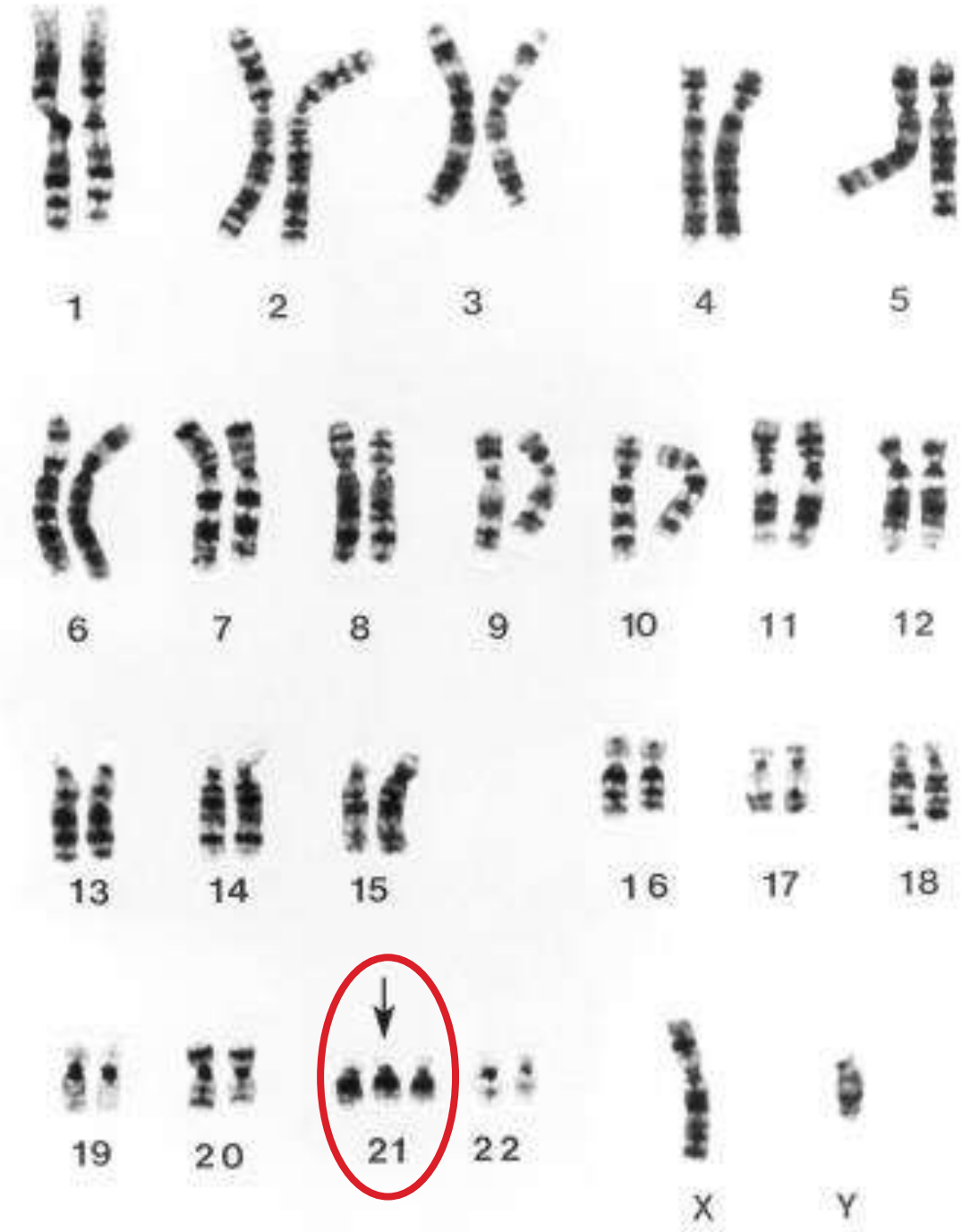
X
Y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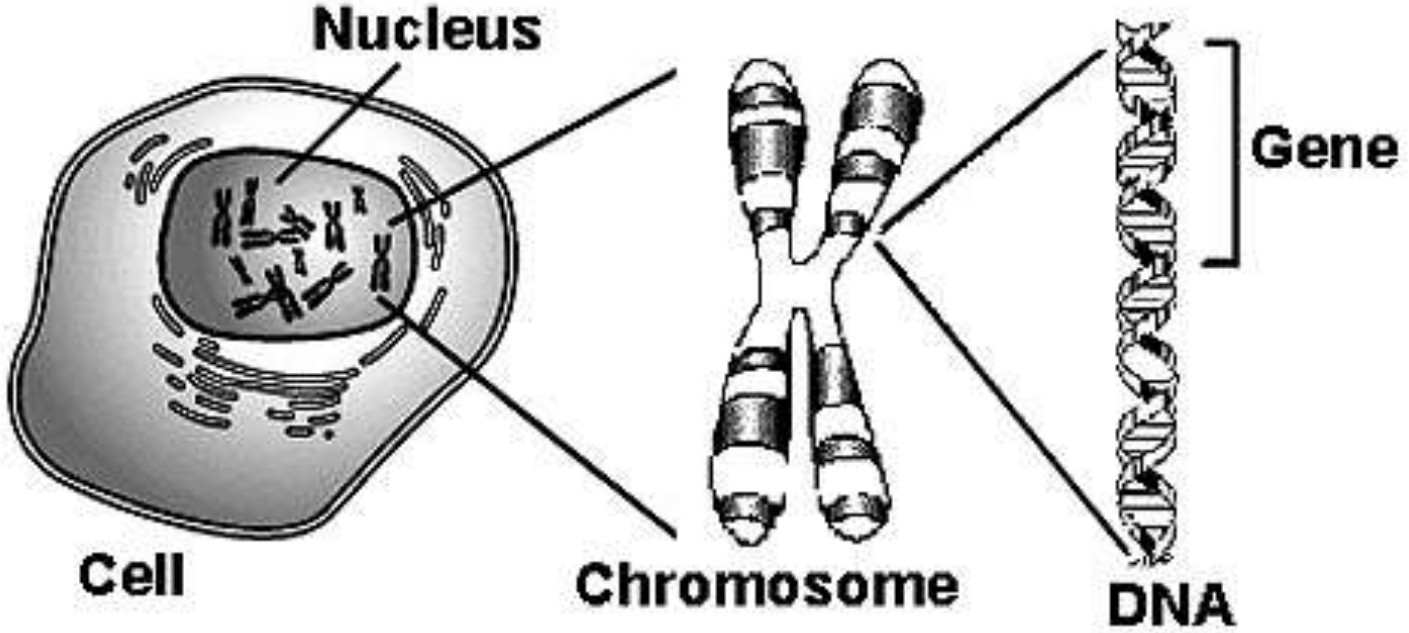
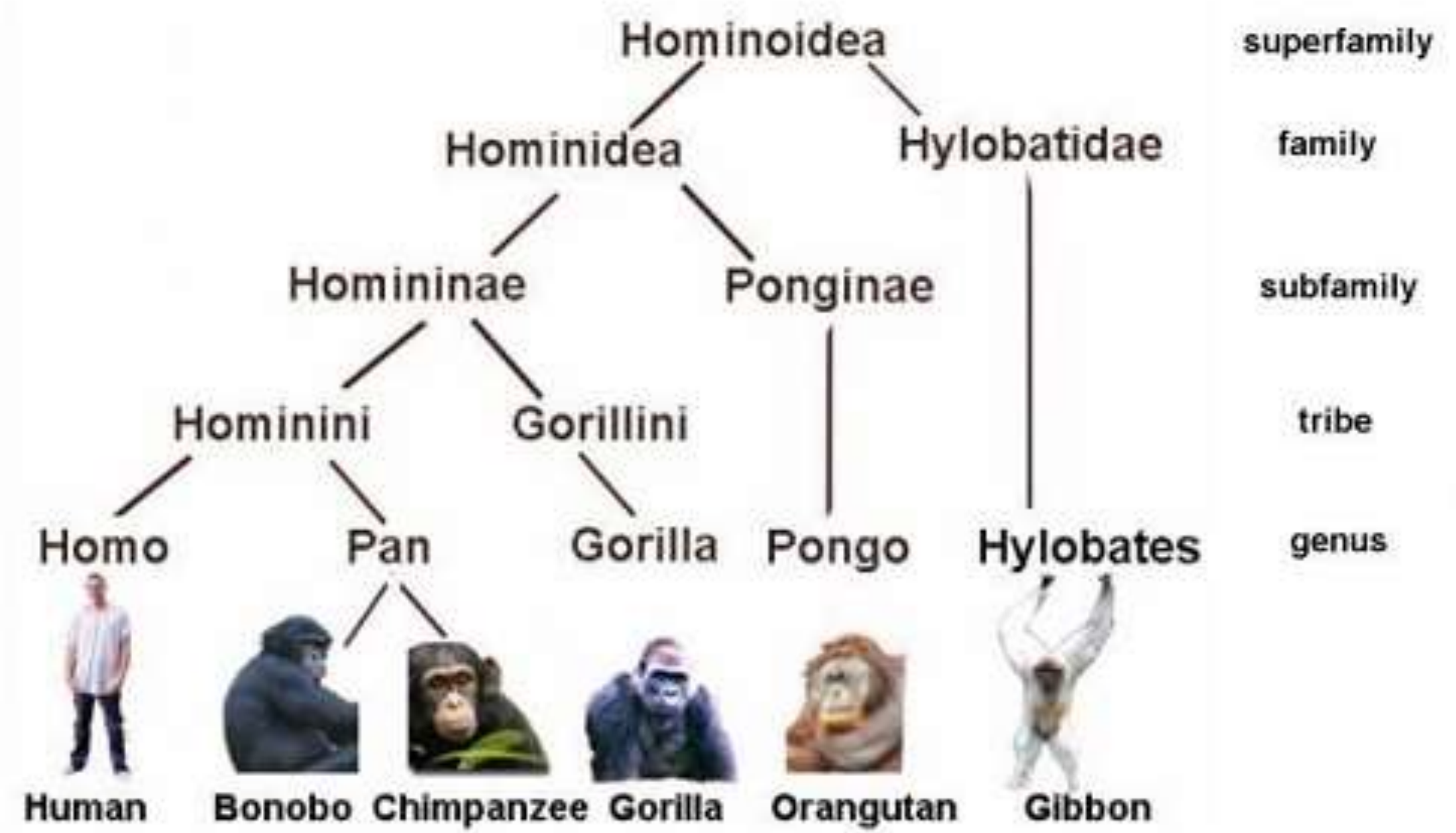

Nucleus
Cell
Chromosome
DNA
Gene

BRANCHES of GENETICS

4. Evolutionary Genetics
   - genetics change within and between species


Hominoidea
superfamily
Hominidea
Hylobatidae
family
Homininae
Ponginae
subfamily
Hominini
Gorillini
tribe
Homo
Pan
Gorilla
Pongo
Hylobates
genus
Human
Bonobo
Chimpanzee
Gorilla
Orangutan
Gibbon

BRANCHES of GENETICS

5. Biochemical Genetics
   - role of enzymes (proteins)
   - products of genes
   - e.g. albinism (homozygous recessive)
     - enzyme? tyrosinase (melanine production)

BRANCHES of GENETICS

6. Behavioral Genetics
   - behavioral traits that are inherited
   - e.g.
     - Schizophrenia
     - alcoholism
     - manic depressive psychosis (excessive happiness and loneliness)
     - criminality

SCHIZOPHRENIA

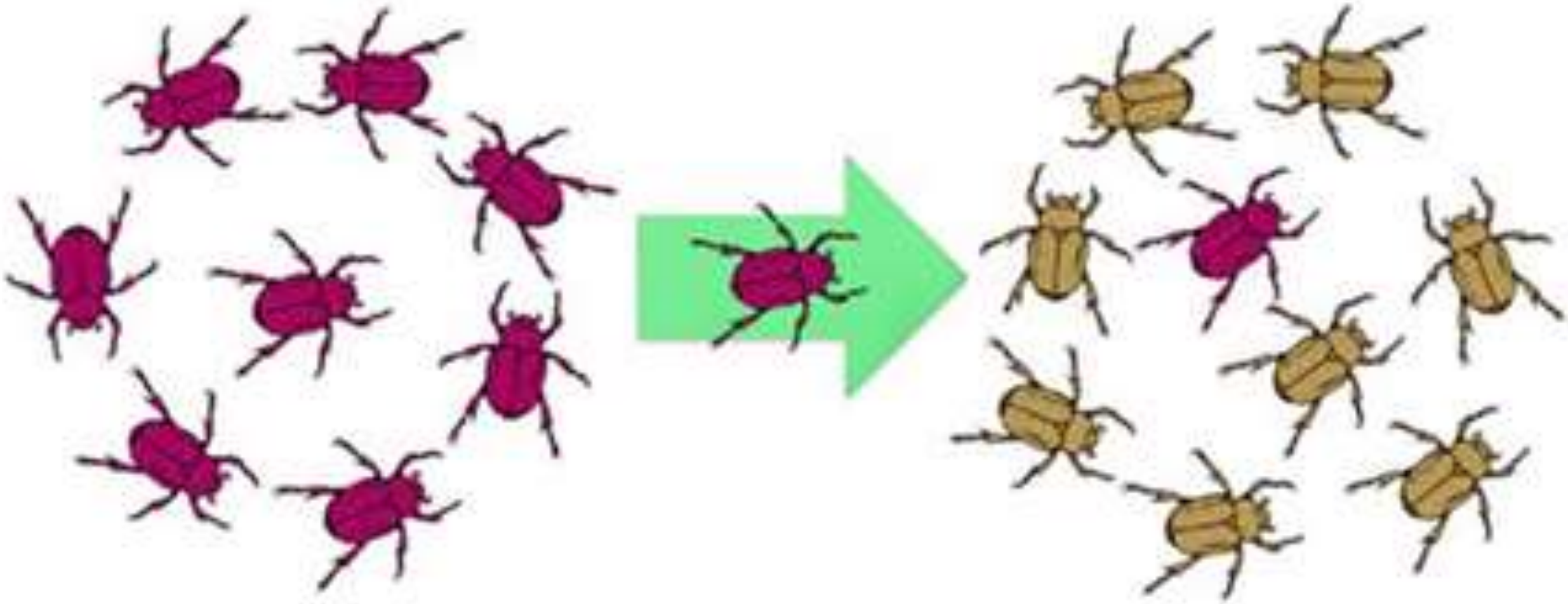
Depressive Symptoms	Manic Symptoms
Gloomy	Elation
Withdrawn	Euphoria
Inability to make decisions	Desire for action
Tired	Hyperactive
Slowness of thought	Multiple ideas

BRANCHES of GENETICS

7. Population Genetics

- fate of genes in the population
- factors affecting gene frequencies
  - mutation
  - selection
  - migration
  - genetic drift

GENE FLOW



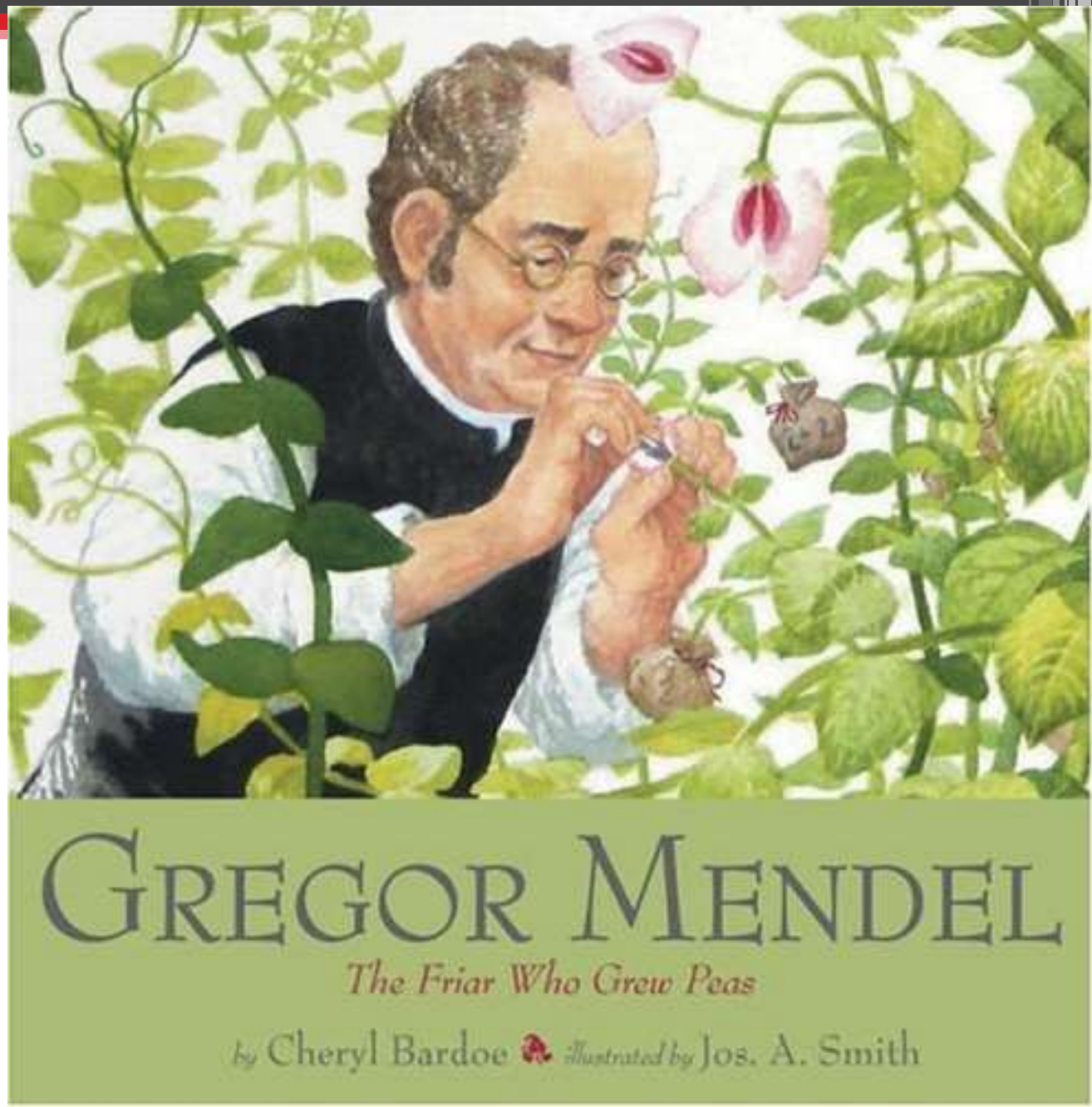
BRANCHES of GENETICS

8. Quantitative Genetics
   - role of genetics (measurable phenotype) and environmental factors in the inheritance and expression of quantitative traits
     - e.g. IQ (proper nutrition, health condition, type of education)

BEGINNINGS of GENETICS

Before Gregor Mendel

- heredity as blending process
  - offsprings are intermediate between the parents
  - not true if offspring resemble either one parent

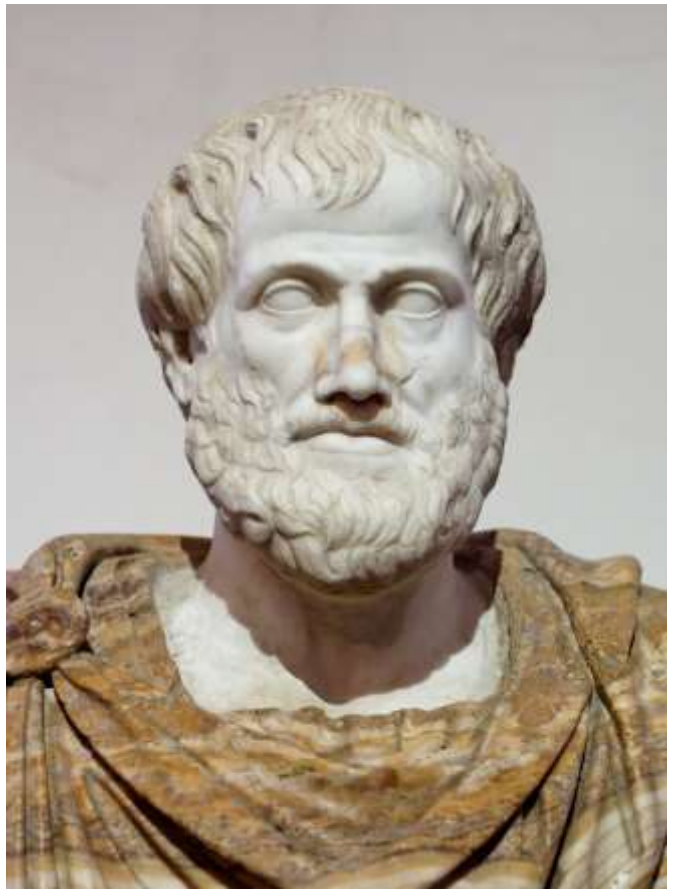

GREGOR MENDEL
The Friar Who Grew Peas
by Cheryl Bardoe
illustrated by Jos. A. Smith

BEGINNINGS of GENETICS

Golden Age of Greek Culture

- attention was given to:
  - reproduction
  - heredity

BEGINNINGS of GENETICS



Aristotle

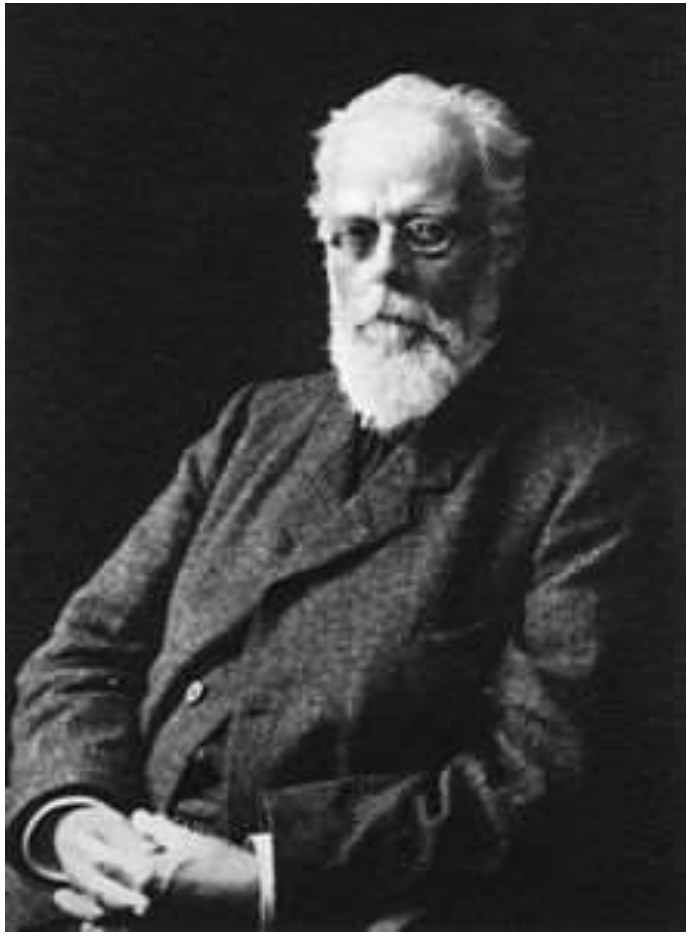
- Theory of Pangenesis
  - all structures and organs of the body contribute copies of themselves to sex cells

BEGINNINGS of GENETICS

Jean Baptiste de Lamarck

- Theory of Inheritance of Acquired Characteristics
  - acquired body modifications are inherited

BEGINNINGS of GENETICS



August Weisman

- Germplasm Theory

(opposite of Pangenesis)

  - plans of the entire body are contributed only by the sex cells

BEGINNING of CLASSICAL GENETICS

Gregor Mendel (1865)

- concept of gene
- presence of discrete hereditary unit
- explains similarities and differences among parents and offsprings

BEGINNING of CLASSICAL GENETICS

Gregor Mendel (1865)

1. He used appropriate experimental tests
   - sell pollinated (*Pisum sativum)*
   - pure line parentals
   - quantitative approach
2. He formulated the theories of segregation and independent assortment

BEGINNING of CLASSICAL GENETICS

Mendel's important contribution to Genetics

1. Appearance of different characteristics in heredity followed specific laws
2. Something from the parent is transmitted to the offspring
3. There is no blending or dilution of inheritance
4. Hereditary units remain unchanged in the presence of other units

BEGINNING of CLASSICAL GENETICS

Carl Correns (German)

Erick Von Tschermak (Austrian)

Hugo de Vries (Netherlands)

- duplicated Mendel's experiments using other plants
- rediscoverers of the work of Mendel

BEGINNING of CLASSICAL GENETICS

William Bateson, Saunders, Lucien Cuenot

- observed Mendel's principles using animals

BEGINNING of CLASSICAL GENETICS

Walter Suttom (USA) and Theodor Boveri (Germany)

- Chromosome Theory of Inheritance

BEGINNING of CLASSICAL GENETICS

Thomas Hunt Morgan and Calvin B. Bridges

- association between specific gene and specific chromosome

BEGINNING of CLASSICAL GENETICS

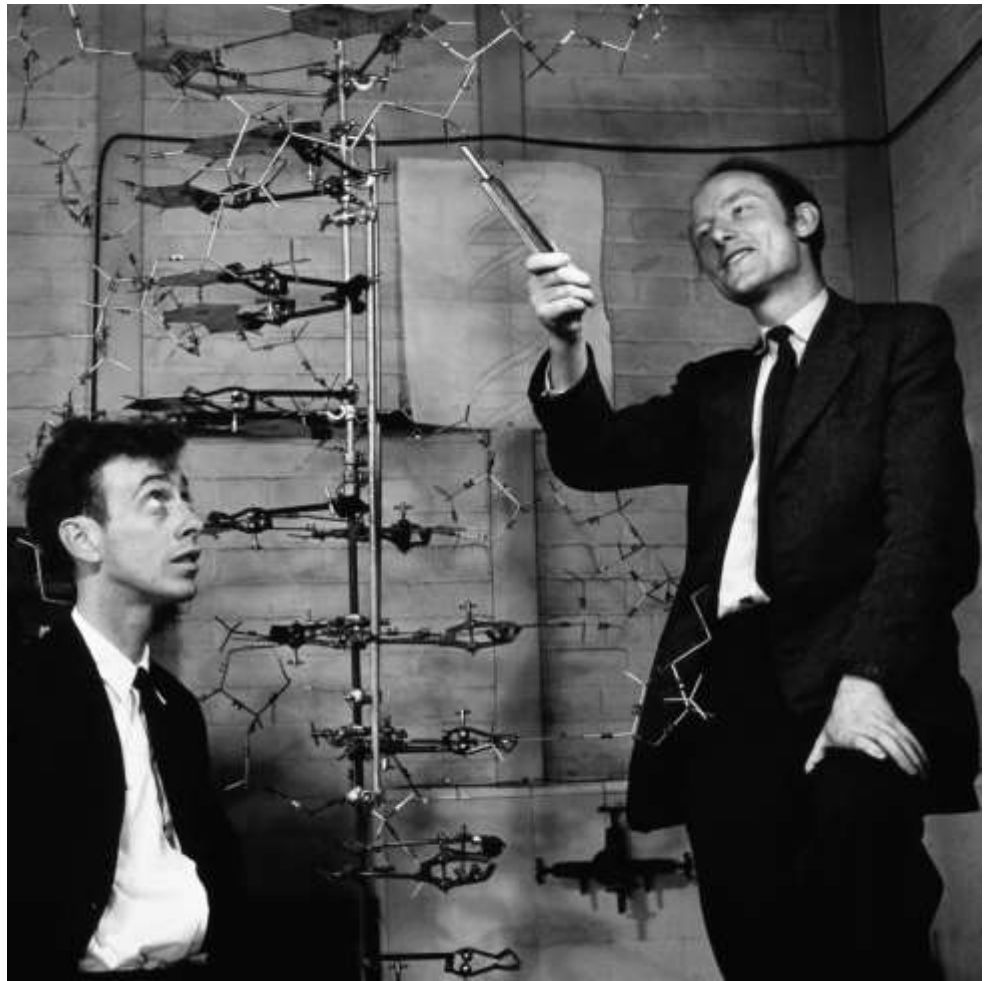
Oswald Avery, Collin Macleod, Maclyn McCarty

- DNA as hereditary material

BEGINNING of CLASSICAL GENETICS

Francis Crick, James Watson, Maurice Wilkins and Rosalind Franklin

- elucidation of the structure of DNA



APPLICATION of GENETICS

1. Microbial, Plant and Animal Improvement
2. Medicine
3. Genetic Counselling
4. Legal Applications
5. Genetic Engineering or Recombinant DNA Technology

APPLICATION of GENETICS

1. Microbial, Plant and Animal Improvement
    - high yielding
    - high nutritional value
    - disease and pest resistance
    - selective breeding in animals
    - carcass quality improvement
    - increase in egg and milk production
    - e.g. native chicken vs improved breeds

(50 eggs/yr vs 300 eggs/yr)

Bt corn
Non-Bt corn
https://www.apsnet.org

APPLICATION of GENETICS

2. Medicine
    - identification of diseases and abnormalities (w/ genetic basis)
    - e.g. metabolic disorders
        - Phenylketonuria
        - Galactosemia

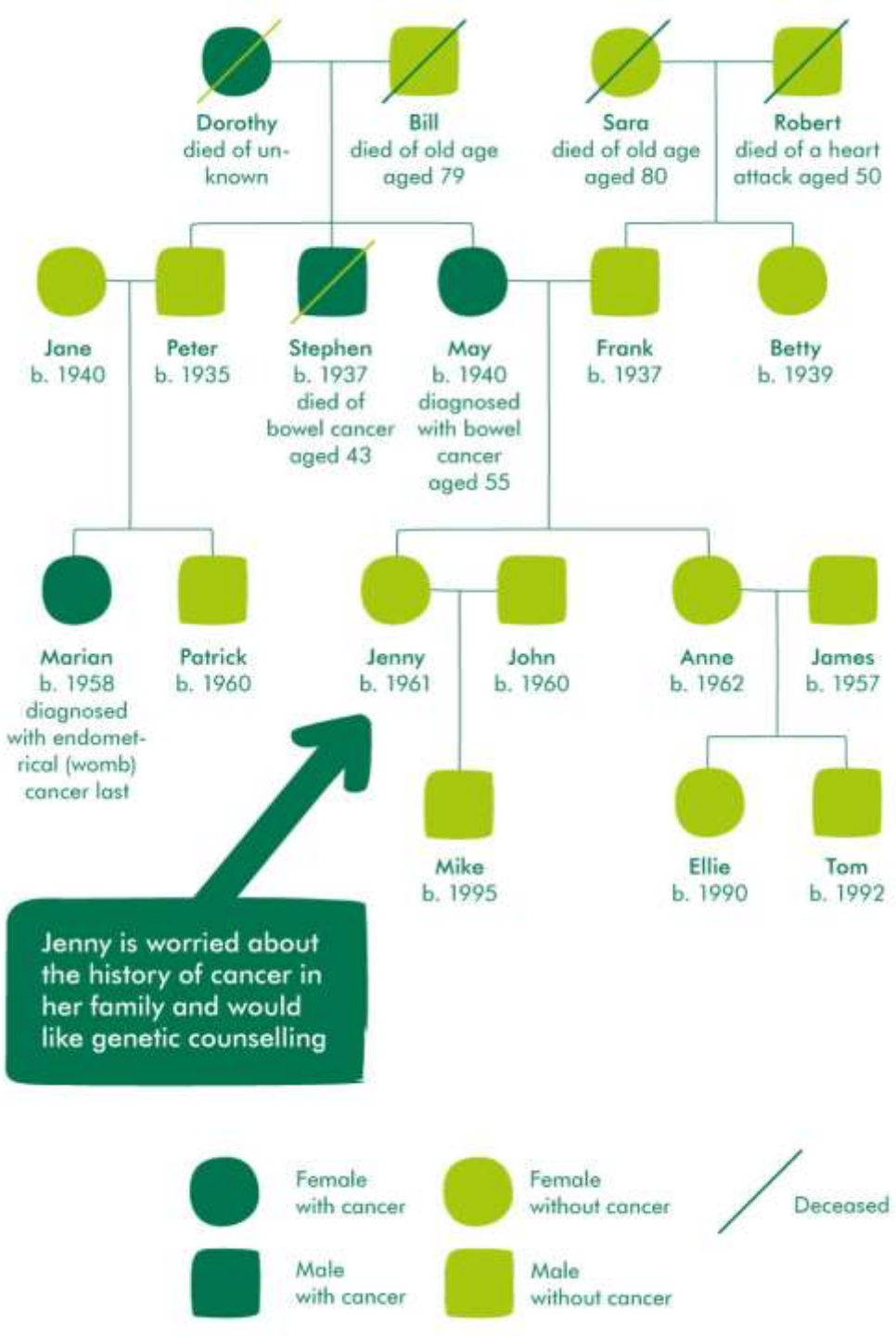
*importance of new born screening

Brain damage
Cataracts
Jaundice
Enlarged liver
Kidney damage
If a galactosemic infant is given milk, unmetabolized milk sugars build up and damage the liver, eyes, kidneys and brain

APPLICATION of GENETICS

3. Genetic Counselling
   - inheritance of undesirable traits
   - pedigree analysis of prospective parents

     - Eugenics
       - coined by Francis Galton in1883 in England
       - application of genetic knowledge for the improvement of human race


Dorothy
died of un-
known
Bill
died of old age
aged 79
Sara
died of old age
aged 80
Robert
died of a heart
attack aged 50
Jane
b. 1940
Peter
b. 1935
Stephen
b. 1937
died of
bowel cancer
aged 43
May
b. 1940
diagnosed
with bowel
cancer
aged 55
Frank
b. 1937
Betty
b. 1939
Marian
b. 1958
diagnosed
with endomet-
rical (womb)
cancer last
Patrick
b. 1960
Jenny
b. 1961
John
b. 1960
Anne
b. 1962
James
b. 1957
Mike
b. 1995
Ellie
b. 1990
Tom
b. 1992
Jenny is worried about
the history of cancer in
her family and would
like genetic counselling
Female
with cancer
Female
without cancer
Deceased
Male
with cancer
Male
without cancer

APPLICATION of GENETICS

3. Genetic Counselling

- Positive Eugenics
  - superior intelligence, intellectual achievers, artists can have large families
- Negative Eugenics
  - mentally retarded and low intelligence are restricted for reproduction

APPLICATION of GENETICS

3. Genetic Counselling

- Negative Eugenics
  - mentally retarded and low intelligence are restricted for reproduction

- Started in Indiana (1907)
  - over half of the states in America passed a law that requires sterilization for imbeciles, idiots, convicted, rapists, habitual criminals

APPLICATION of GENETICS

3. Genetic Counselling

- Negative Eugenics
  - mentally retarded and low intelligence are restricted for reproduction
- 1931 involuntary sterilization
  - sexual perverts, drug addicts, drunkards, epileptics

*immigration to US is restricted because certain areas in Europe and Asia are genetically inferior

*Adolf Hitler (1930)

- forced sterilization
- mercy killing (genocide?)

APPLICATION of GENETICS

3. Genetic Counselling

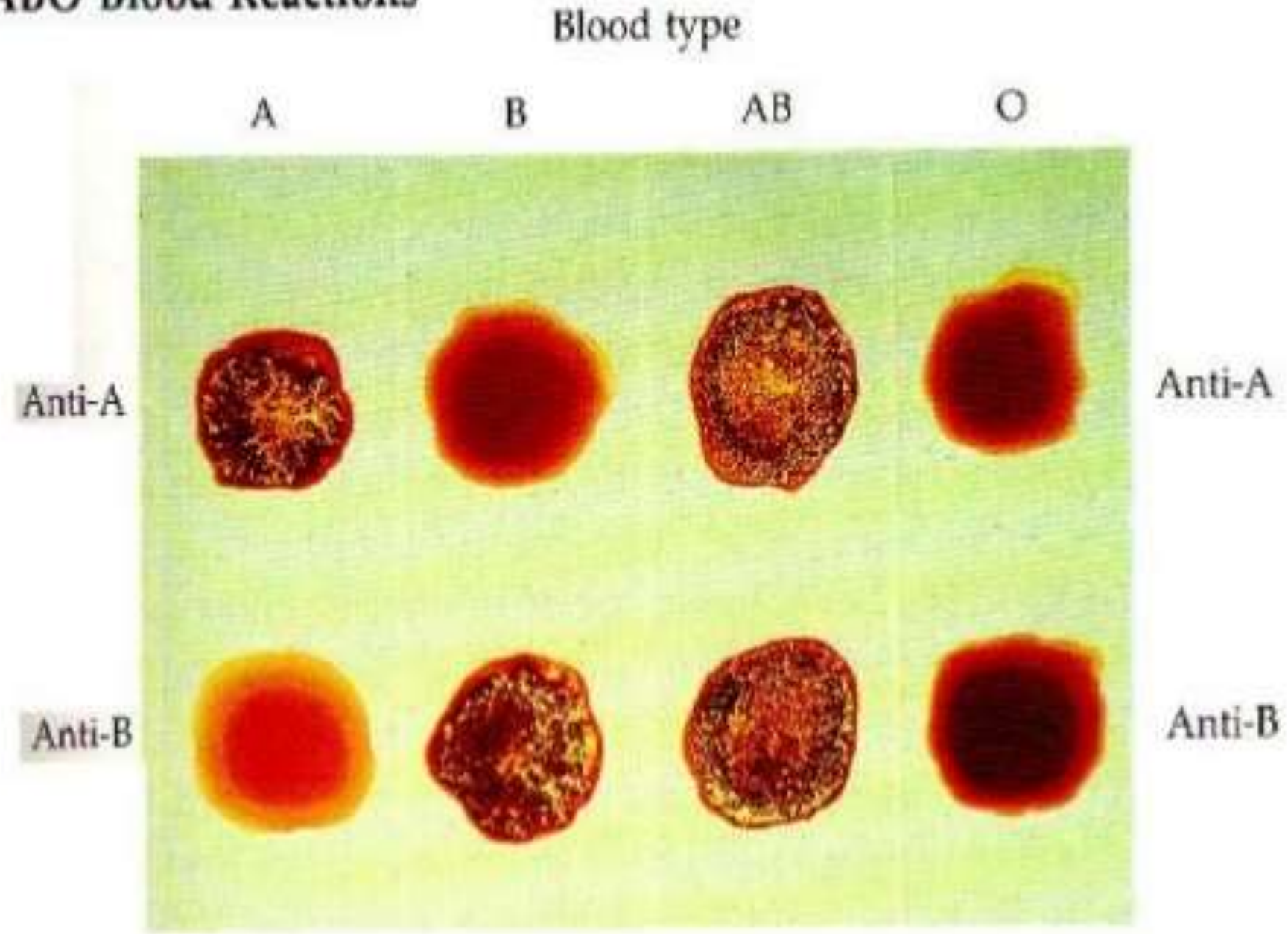
- Euphenics
  - replaced Eugenics
  - medical or genetic intervention designed to reduce impact of defective genotypes or individuals
  - e.g.
    - insulin for diabetes
    - dietary control (controlling galactose in galactosemic individuals)
    - genetic surgery (future?)

APPLICATION of GENETICS

4. Legal Applications

- blood type analysis
- DNA fingerprinting
  - paternity testing
  - disputed parentage
  - identify criminals

ABO Blood Reactions

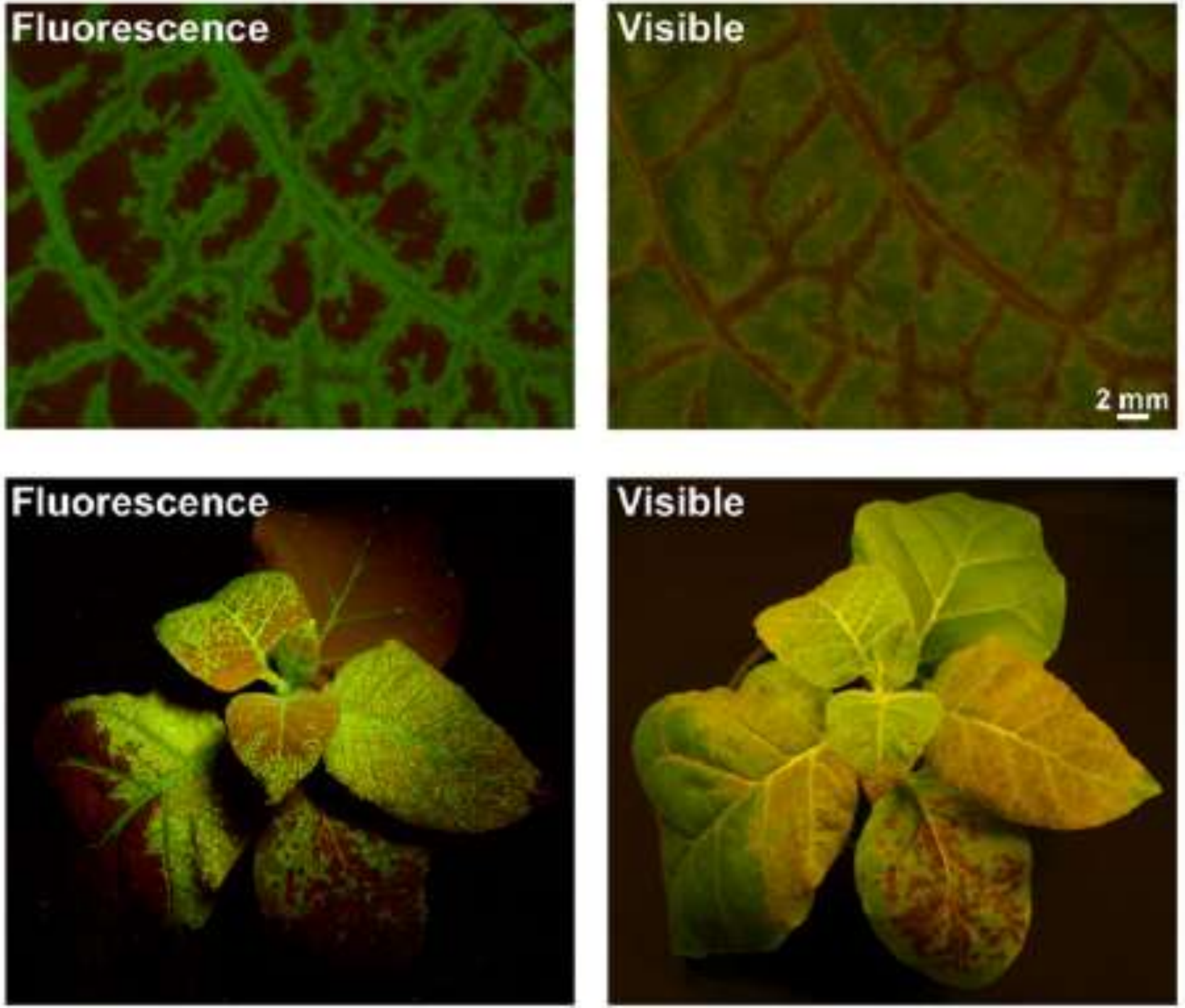


APPLICATION of GENETICS

5. Genetic Engineering or Recombinant DNA Technology

- GMO
  - Bt corn, transgenic papaya, cotton, soybean, etc.
- products of recombinant DNA technology
  - humulin
  - transgenic tobacco w/ luciferase gene
  - Golden rice rich in pro Vitamin A

Humulin R
REGULAR
insulin human
injection, USP
(rDNA origin)
U-100
Humulin R
U-100
REGULAR
insulin human injection, USP (rDNA origin)


Fluorescence
Visible
2 mm
Fluorescence
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